Letter to the Editor

Multiple Vertebral Segmentation Defects and Rib Anomalies

To the Editor:

I have read with great interest the paper by Mortier et al. [1996] on multiple vertebral segmentation defects. These authors define three distinct types of short trunk dwarfism with multiple vertebral segmentation defects and rib anomalies. The diagnosis of the first entity (the so-called Jarcho-Levin syndrome) is based mainly on its radiographic characteristics of "crab-like" thorax and the suggestion of autosomal recessive inheritance. The diagnosis of the second entity (frequently called spondylothoracic dysostosis) is essentially based on its autosomal recessive inheritance, and the third one (considered as spondylocostal dysostosis) on its autosomal dominant mode of inheritance. Mortier et al. [1996] concluded that spondylocostal dysostosis shows a considerable clinical and radiographic overlap with spondylothoracic dysostosis. These authors also consider that other major malformations are uncommon in these 3 entities. They also consider one more group constituted by the sporadic patients, which is quite heterogeneous and more frequently associated with other congenital anomalies. Thus, the differences between the entities defined by Mortier et al. [1996] are partially based on the clinical and radiographic findings and partially on the genetic aspects.

In 1994 we published an epidemiological analysis of segmentation anomalies of the vertebrae and ribs [Martínez-Frías and Urioste, 1994] in which we demonstrated that all degrees of severity in vertebral and rib anomalies were observed in different multiple congenital anomaly (MCA) patterns and causally identified conditions such as monogenic syndromes, aneuploidy syndromes, or after prenatal exposure to environmental agents, and we concluded that segmentation anomalies of the axial skeleton constitute a dysmorphogenetic reaction of the primary developmental field, and as such, a developmental field defect (DFD). Thus, the radiographic characteristics are not a good criterion to classify the distinct types of short trunk dwarfism, because the anomalies of the axial skeleton, as expected in a DFD, could be observed in different degrees of severity in different clinical and causal conditions. Moreover, in a previous paper [Martínez-Frías et al., 1994] we identified epidemiologically that among infants with nonsyndromal MCA patterns, the different degrees of vertebrae and rib anomalies tend to be preferentially associated with central nervous system defects, including neural tube defects, with the caudal dysgenesis complex, and with diaphragmatic hernia. These results suggest the existence of other different entities that should be investigated further.

In conclusion, I think that considering the different anomalies of the axial skeleton as a primary DFD clarifies the great variability in their clinical and radiographic manifestation in different causally identified conditions and nonsyndromal MCA patterns. Consequently, the cases should be classified on the basic cause and, for those with a MCA pattern, taking into account a possible common pathogenetic mechanism, rather than by radiographic characteristics.

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